

Title (en)
METHOD TO DETECT REPEAT SEQUENCE MOTIFS IN NUCLEIC ACID

Title (de)
VERFAHREN ZUR ERKENNUNG VON WIEDERHOLUNGSSEQUENZMOTIVEN IN NUKLEINSÄURE

Title (fr)
PROCÉDÉ DE DÉTECTION DE MOTIFS DE SÉQUENCES RÉPÉTÉES DANS UN ACIDE NUCLÉIQUE

Publication
EP 2531618 A4 20130925 (EN)

Application
EP 11740443 A 20110204

Priority
• US 30205910 P 20100205
• US 2011023777 W 20110204

Abstract (en)
[origin: WO2011097503A2] Methods for determining the presence or absence of expansion of CGG repeat sequence in the FMR1 gene presence or absence of expansion of CCG repeat sequence in the FMR2 gene are provided. The methods are useful in identifying an individual with normal/intermediate, versus premutation or full mutation allele of FMR1 gene and FMR2 gene due to the expansion of CGG repeats and CCG repeats in the 5'-untranslated region respectively. The methods are also useful for screening newborns for fragile X syndrome or for screening women to determine heterozygosity status with full premutation of the CCG repeat tract. The methods are also useful in estimating the premutation and full mutation carrier frequency and estimating the prevalence of FXTAS AND FXPOI in a population. The methods are simple, rapid and require small amount of sample.

IPC 8 full level
C12Q 1/68 (2006.01); **C12P 19/34** (2006.01); **G16B 25/20** (2019.01)

CPC (source: EP US)
C12Q 1/6827 (2013.01 - EP US); **C12Q 1/683** (2013.01 - EP US); **C12Q 1/6858** (2013.01 - EP US); **C12Q 1/6883** (2013.01 - EP US); **G16B 25/20** (2019.01 - EP US); **C12Q 2600/154** (2013.01 - EP US); **C12Q 2600/156** (2013.01 - EP US); **C12Q 2600/158** (2013.01 - US); **C12Q 2600/16** (2013.01 - EP US); **G16B 99/00** (2019.01 - EP US)

Citation (search report)
• [XP] WO 2010110835 A1 20100930 - ASURAGEN INC [US], et al
• [I] TASSONE F ET AL: "A rapid polymerase chain reaction-based screening method for identification of all expanded alleles of the fragile X (FMR1) gene in newborn and high-risk populations", JOURNAL OF MOLECULAR DIAGNOSTICS, THE, AMERICAN SOCIETY FOR INVESTIGATIVE PATHOLOGY, US, vol. 10, no. 1, 1 January 2008 (2008-01-01), pages 43 - 49, XP002592250, ISSN: 1525-1578, [retrieved on 20071228], DOI: 10.2353/JMOLDX.2008.070073
• See references of WO 2011097503A2

Designated contracting state (EPC)
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US 2011023777 W 20110204; EP 11740443 A 20110204; EP 16151944 A 20110204; EP 18188881 A 20110204; EP 20200239 A 20110204; EP 23195085 A 20110204; US 201113576740 A 20110204; US 201615174038 A 20160606; US 201916357247 A 20190318; US 202217963045 A 20221010